

Table 1 : Comparison of diagnostic methods for SMA

| N° tube        | SSCP <sup>(1)</sup> | Our méthode (radioactive) <sup>(3)</sup> |                     |                      |                  |                     |                      |
|----------------|---------------------|--|---------------------|----------------------|------------------|---------------------|----------------------|
|                |                     | Probe 1 (exon 7)                         |                     |                      | Probe 2 (exon 8) |                     |                      |
|                |                     | Exons <sup>(2)</sup>                     | PSL/mm <sup>2</sup> | R (%) <sup>(4)</sup> | Exon 7           | PSL/mm <sup>2</sup> | R (%) <sup>(4)</sup> |
| Control<br>(1) | ndel 7/ndel 8       | 42                                       | 0                   | ndel                 | 22               | 0                   | ndel                 |
| Control<br>(2) | ndel 7/ndel 8       | 41                                       | 0                   | ndel                 | 23               | 0                   | ndel                 |
| Control<br>(3) | ndel 7/ndel 8       | 43                                       | 0                   | ndel                 | 22               | 0                   | ndel                 |
| Control<br>(4) | ndel 7/ndel 8       | 41                                       | 0                   | ndel                 | 21               | 0                   | ndel                 |
| Control<br>(5) | ndel 7/ndel 8       | 42                                       | 0                   | ndel                 | 23               | 0                   | ndel                 |
| SMA<br>(6)     | del 7/del 8         | 24                                       | 43                  | del                  | 15               | 32                  | del                  |
| SMA<br>(7)     | del 7/del 8         | 14                                       | 67                  | del                  | 08               | 64                  | del                  |
| SMA<br>(8)     | del 7/del 8         | 10                                       | 76                  | del                  | 07               | 68                  | del                  |
| SMA<br>(9)     | del 7/del 8         | 26                                       | 38                  | del                  | 06               | 73                  | del                  |
| SMA<br>(10)    | del 7/del 8         | 09                                       | 79                  | del                  | 13               | 41                  | del                  |
| SMA<br>(11)    | del 7/del 8         | 27                                       | 36                  | del                  | 12               | 45                  | del                  |
| SMA<br>(12)    | del 7/del 8         | 15                                       | 64                  | del                  | 15               | 32                  | del                  |
| SMA<br>(13)    | del 7/del 8         | 13                                       | 69                  | del                  | 14               | 36                  | del                  |
| SMA<br>(14)    | del 7/del 8         | 25                                       | 40                  | del                  | 11               | 50                  | del                  |
| SMA<br>(15)    | del 7/del 8         | 20                                       | 52                  | del                  | 09               | 59                  | del                  |
| SMA<br>(16)    | del 7/del 8         | 19                                       | 55                  | del                  | 13               | 41                  | del                  |
| SMA<br>(17)    | del 7/del 8         | 12                                       | 71                  | del                  | 14               | 36                  | del                  |
| SMA<br>(18)    | del 7/del 8         | 14                                       | 67                  | del                  | 12               | 45                  | del                  |

(1) single strand conformation polymorphism

(2) del : deleted ; ndel : non deleted

(3) The quantification of results obtained is performed by means of Bio-Imager and expressed as PSL/mm<sup>2</sup>

(4) R : difference = 1 - [(PSL/mm<sup>2</sup>Control - PSL/mm<sup>2</sup>SMA)/(PSL/mm<sup>2</sup>Control)]

The mean value of the control group is used for the calculation of R

Table 2 : Comparison of diagnostic methods for SMA

| N° tube     | SSCP <sup>(1)</sup> | Our method (ELISA)   |                 |                     |                  |                 |                     |                       |                 |
|-------------|---------------------|----------------------|-----------------|---------------------|------------------|-----------------|---------------------|-----------------------|-----------------|
|             |                     | Probe 1 (exon 7)     |                 |                     | Probe 2 (exon 8) |                 |                     | Probe 3<br>(HUMEF1AB) |                 |
|             |                     | Exons <sup>(2)</sup> | Optical density | R(%) <sup>(3)</sup> | Exon 7           | Optical density | R(%) <sup>(3)</sup> | Exon 8                | Optical density |
| Control (1) | ndel 7/<br>ndel 8   | 0.26                 | 0               |                     | ndel             | 0.28            | 0                   | ndel                  | 0.55            |
| Control (2) | ndel 7/<br>ndel 8   | 0.28                 | 0               |                     | ndel             | 0.29            | 0                   | ndel                  | 0.54            |
| Control (3) | ndel 7/<br>ndel 8   | 0.27                 | 0               |                     | ndel             | 0.26            | 0                   | ndel                  | 0.52            |
| Control (4) | ndel 7/<br>ndel 8   | 0.26                 | 0               |                     | ndel             | 0.28            | 0                   | ndel                  | 0.51            |
| Control (5) | ndel 7/<br>ndel 8   | 0.27                 | 0               |                     | ndel             | 0.27            | 0                   | ndel                  | 0.49            |
| SMA (6)     | del 7/<br>del 8     | 0.16                 | 41              |                     | del              | 0.13            | 54                  | del                   | 0.51            |
| SMA (7)     | del 7/<br>del 8     | 0.16                 | 41              |                     | del              | 0.16            | 43                  | del                   | 0.48            |
| SMA (8)     | del 7/<br>del 8     | 0.11                 | 59              |                     | del              | 0.12            | 57                  | del                   | 0.53            |
| SMA (9)     | del 7/<br>del 8     | 0.15                 | 44              |                     | del              | 0.14            | 50                  | del                   | 0.49            |
| SMA (10)    | del 7/<br>del 8     | 0.19                 | 30              |                     | del              | 0.16            | 43                  | del                   | 0.5             |
| SMA (11)    | del 7/<br>del 8     | 0.13                 | 52              |                     | del              | 0.12            | 57                  | del                   | 0.49            |
| SMA (12)    | del 7/<br>del 8     | 0.14                 | 48              |                     | del              | 0.13            | 54                  | del                   | 0.48            |
| SMA (13)    | del 7/<br>del 8     | 0.17                 | 37              |                     | del              | 0.14            | 50                  | del                   | 0.55            |
| SMA (14)    | del 7/<br>del 8     | 0.12                 | 55              |                     | del              | 0.15            | 46                  | del                   | 0.52            |
| SMA (15)    | del 7/<br>del 8     | 0.11                 | 59              |                     | del              | 0.13            | 54                  | del                   | 0.5             |
| SMA (16)    | del 7/<br>del 8     | 0.13                 | 52              |                     | del              | 0.15            | 46                  | del                   | 0.51            |
| SMA (17)    | del 7/<br>del 8     | 0.15                 | 44              |                     | del              | 0.12            | 57                  | del                   | 0.49            |
| SMA (18)    | del 7/<br>del 8     | 0.16                 | 41              |                     | del              | 0.16            | 43                  | del                   | 0.5             |

(1) single strand conformation polymorphism

(2) del : deleted ; ndel : non deleted

(3) R : difference =  $1 - [(OD_{450} \text{Control} - OD_{450} \text{SMA}) / OD_{450} \text{Control}]$

The mean value of the control group is used for the calculation of R